



IN THE UNITED STATES PATENT AND TRADEMARK OFFICE

Application of:

KNOLL, JOAN et al.

Serial No. :09/854,867

Filed: May 14, 2001

SINGLE COPY GENOMIC  
HYBRIDIZATION PROBES AND  
METHOD OF GENERATING SAME

Docket No.30307A

Group Art Unit No. 1634

Examiner: C. Myers

Commissioner of Patents  
Alexandria, VA 22313-1450


Sir:

PETITION TO THE COMMISSIONER  
UNDER 37 CFR § 1.84(a)(2) TO ACCEPT PHOTOGRAPHS

Applicant hereby petitions the Commissioner of Patents and Trademarks, under 37 CFR § 1.84(a)(2) to accept these photographs into the above-referenced application. Enclosed herewith are three (3) sets of photographs to be entered into the above-referenced application; along with one set of black and white copies that accurately depicts, to the extent possible, the subject matter shown in the color drawings. A check in the amount of \$130.00 in accordance with § 1.17 (h). Applicant requests favorable consideration of this petition

Any additional fees which are due in connection with this Petition should be applied against our Deposit Account No. 19-0522.

Respectfully submitted,

By   
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12/30/2004 AWONDAF1 00000126 09854867

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ATTORNEYS FOR APPLICANT(S)

**BEST AVAILABLE COPY**

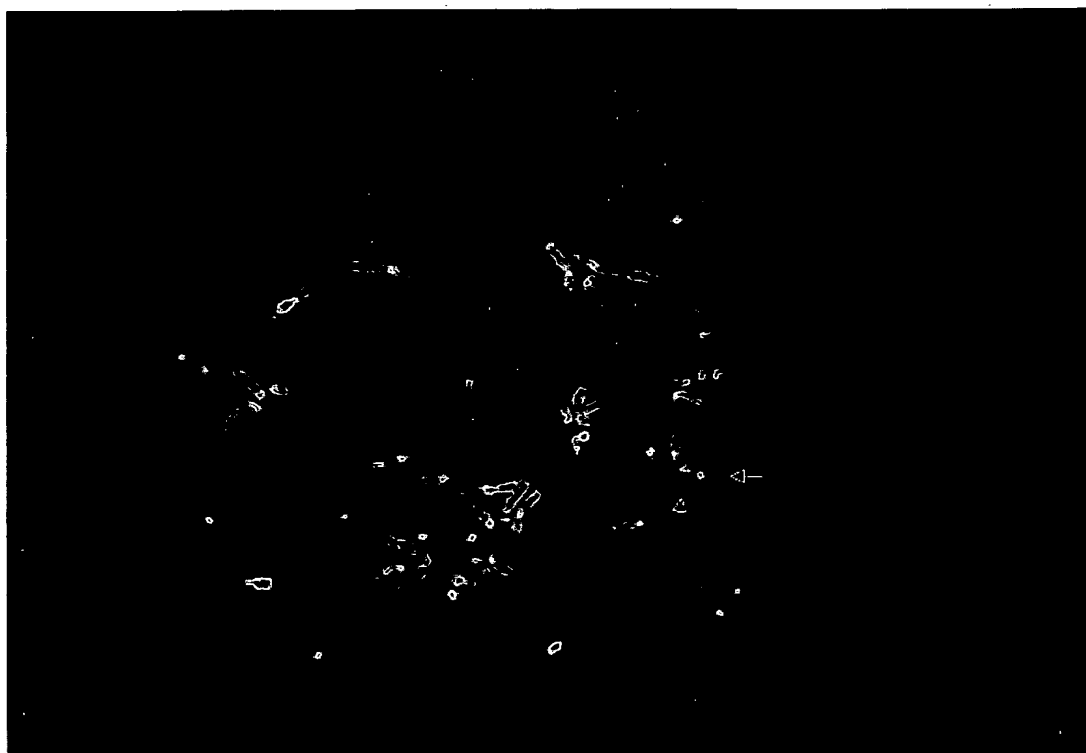


Fig. 1

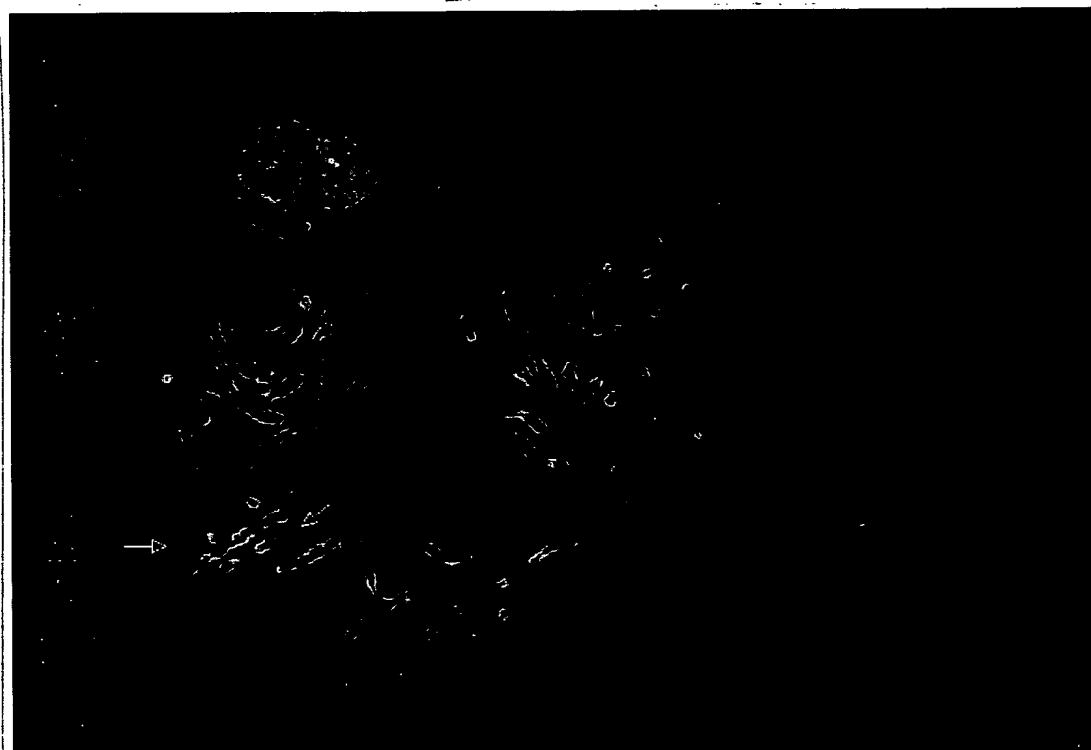


Fig. 2

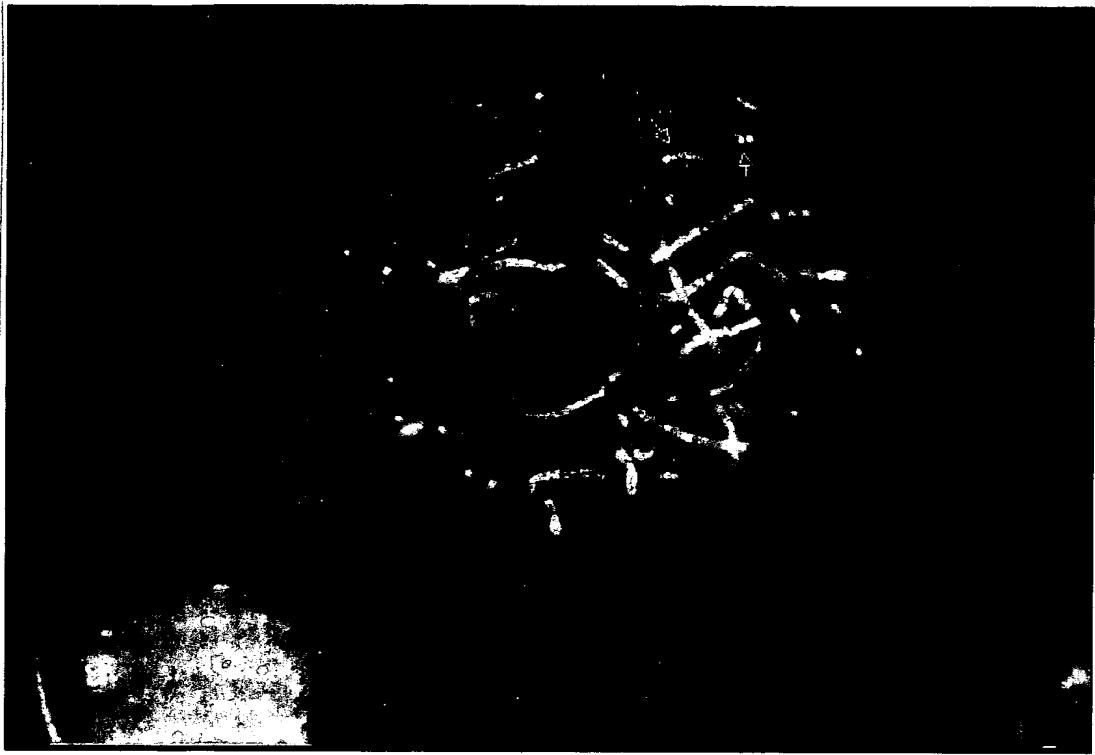


Fig. 3

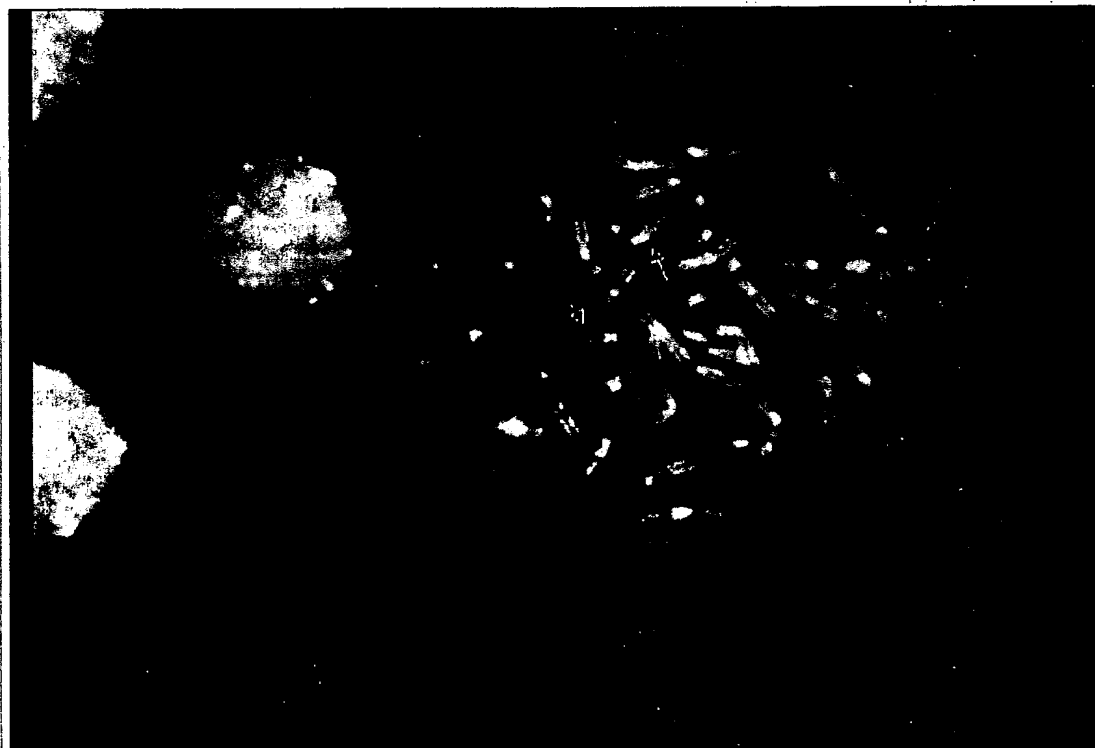


Fig. 4

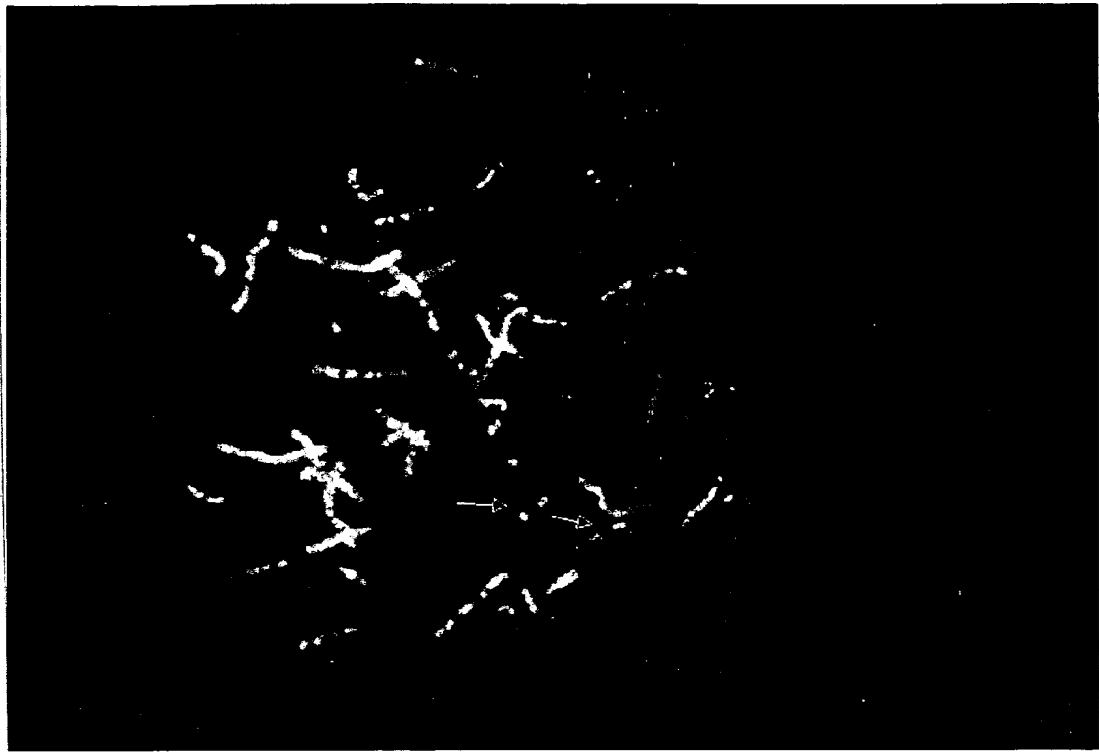


Fig. 5

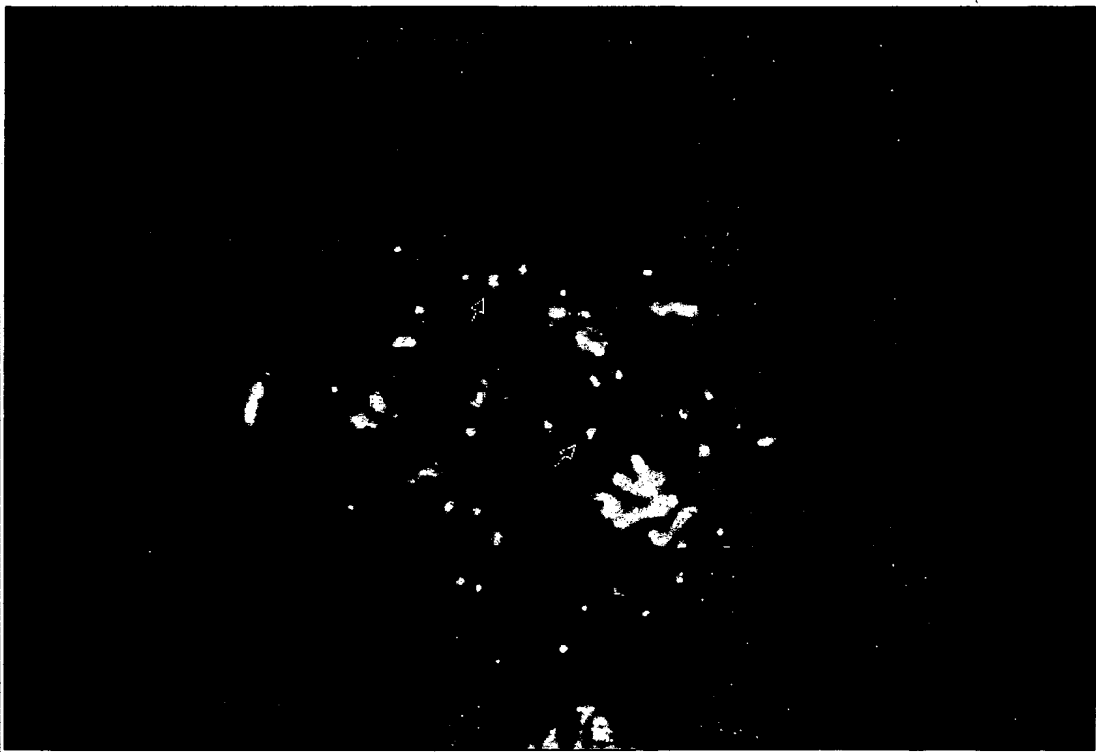


Fig. 6



Fig. 7



Fig. 8



Fig. 9

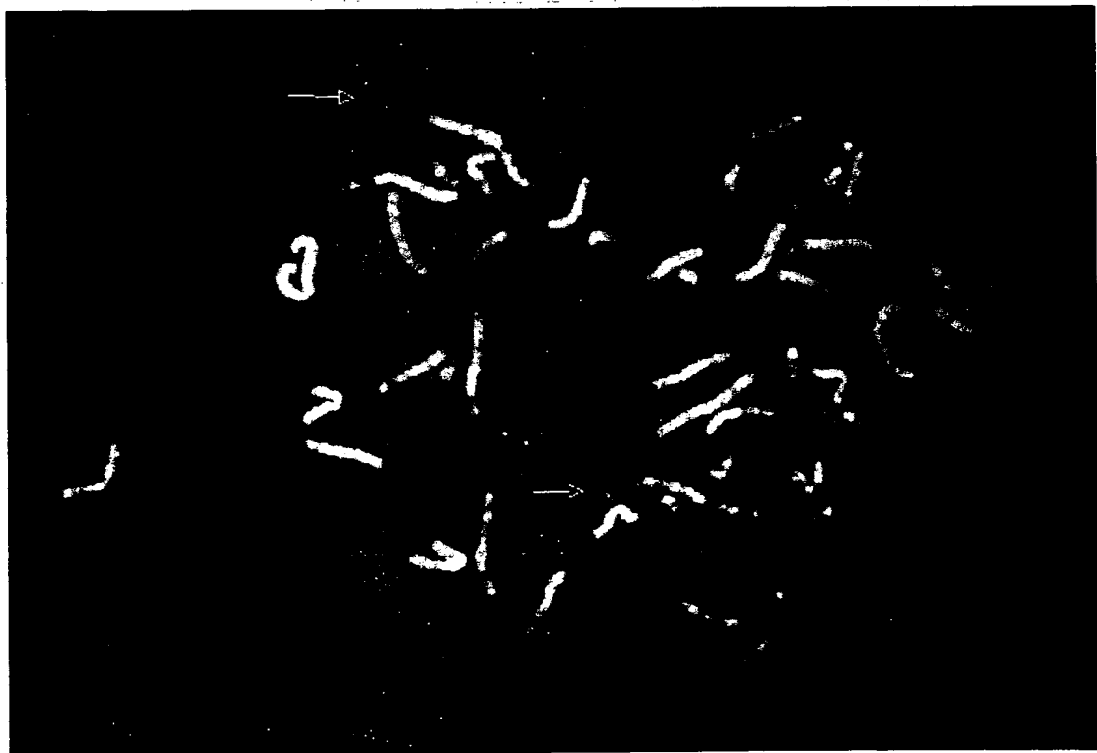


Fig. 10

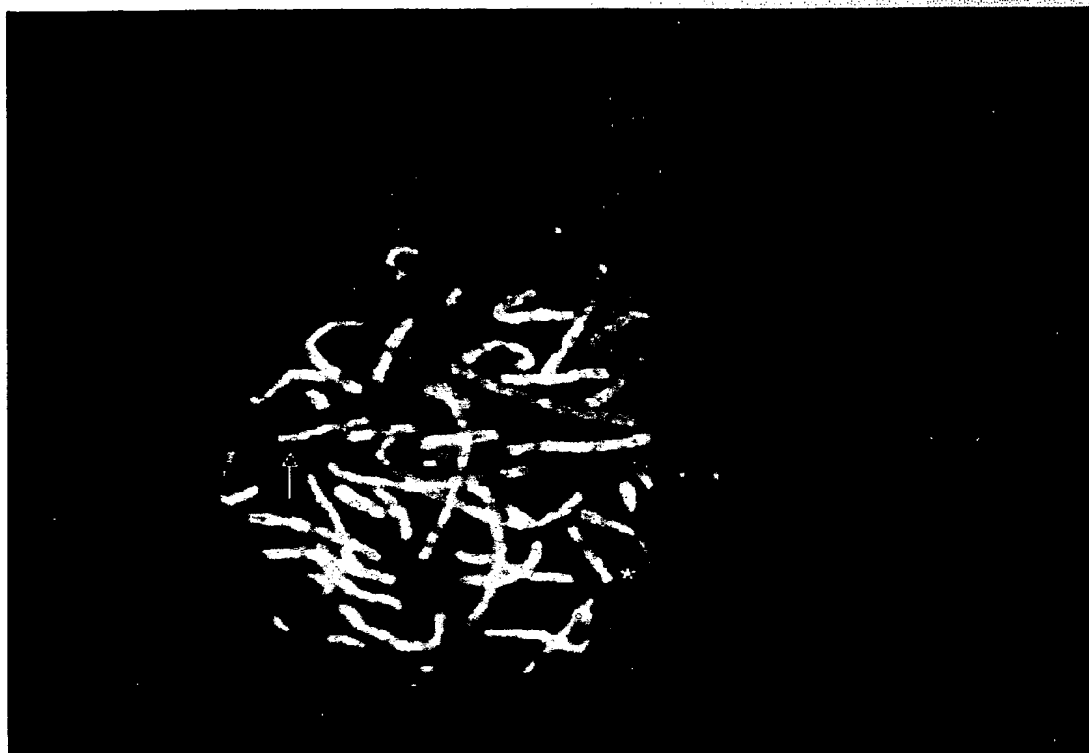
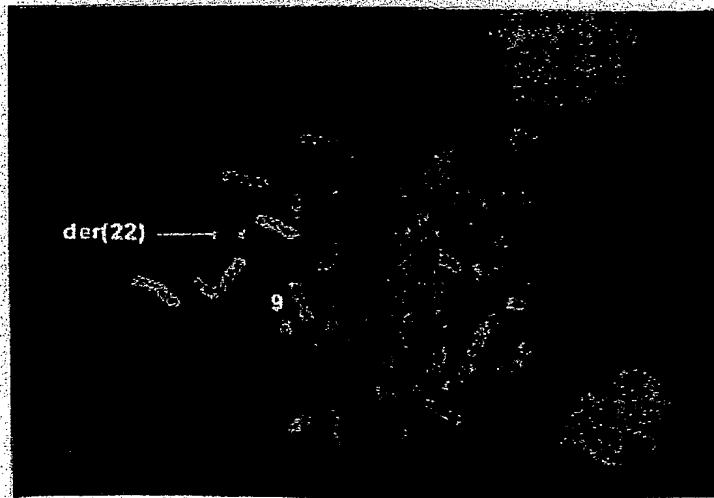


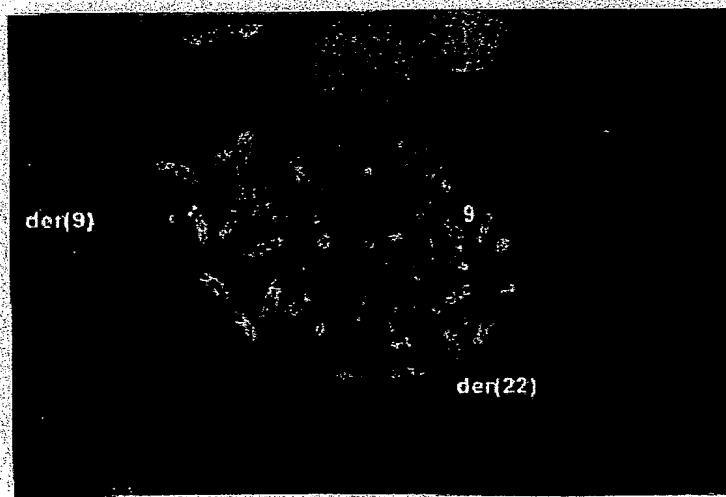
Fig. 11



Fig. 12



**Fig. 15**



**Fig. 16**





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#### EDUCATION

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1979	B.A.	Biophysics	Johns Hopkins University
1980	D.E.A.	Cellular Biology	Université de Grenoble, France
1983	M.Phil.	Molecular Biophysics and Biochemistry	Yale University
1987	Ph.D.	Molecular Biophysics and Biochemistry (Human Genetics)	Yale University (Advisor: S. M. Weissman, M.D.)

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#### WORK EXPERIENCE AND POSTGRADUATE TRAINING

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1979 - 1980	Research Assistant, Biophysics Laboratory	Institut Laue-Langevin Grenoble, France
1984 - 1986	Consultant, Computational Molecular Biology	Dept. Human Genetics, Yale University School of Medicine
1987 - 1990	Postdoctoral Fellow, Laboratory of Eukaryotic Gene Expression	ABL/Basic Research Program National Cancer Institute, NIH
1990 - 1996	Assistant Professor, Dept. Pediatrics, Div. Genetics	College of Medicine, The Pennsylvania State University
1993	Participant, Genetic Linkage Analysis Course (Director, J. Ott)	College of Physicians and Surgeons, Columbia University
1995 - present	President	Phylogenetix Laboratories, Inc.
1996 - 1999	Associate Professor, Human Genetics	MCP Hahnemann School of Medicine
1999 - 2004	Associate Professor, Pediatrics	School of Medicine, University of Missouri-Kansas City
2001 - 2004	Associate Professor, Computer Science	School of Interdisciplinary Computing and Engineering, University of Missouri-Kansas City
2002- present	Adjunct Associate Professor	Department of Chemistry, University of Kansas
2004 - present	Professor, Pediatrics and Computer Science	Schools of Medicine and Computer Science & Engineering, University of Missouri-Kansas City

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#### HONORS, AWARDS AND PROFESSIONAL RECOGNITION

Bourse d'Etudes (Government of France), 1979-1980  
National Research Service Predoctoral Award, 1980-1984  
Postdoctoral Fellowship Award, ABL-Basic Research Program, 1987-1988

Scientific Conference Coordinator: Prader-Willi Syndrome Association (National), 1992  
Basil O'Conner Starter Scholar, March of Dimes, 1992-1993  
Shannon Award, National Institutes of Health, 1992-1993  
Elected to Human Genome Organization, 1992  
Director, March of Dimes-Central PA Molecular Diagnostic Laboratory, 1993-1996  
March of Dimes Birth Defects Foundation Community Service Award, 1994  
Honors, Pennsylvania Society of Professional Engineers -Harrisburg chapter, 1996  
Who's Who in the East, 1997  
Translational Research Award, Children's Cancer Group, 1999  
Professorship in Pediatric Molecular Genetics, Children's Mercy Hospital, UMKC School of Medicine, 1999  
External Advisory Board; Pharmacogenetics of Anticancer Agents (PharmGKB network, NIGMS PHS), 8/2003-.

#### **PROFESSIONAL AFFILIATIONS**

Sigma XI, 1987- ; American Association for the Advancement of Science, 1984- ; American Society of Human Genetics, 1990- ; American Association for Cancer Research, 1996- ; Human Genome Organization, 1992- ; Mutation Database Association 1997-; International Society for Computational Biology 2003-

#### **GRANTS REVIEWED**

Louis B. Leakey Foundation, US; National Science Foundation, US; Action Research Charity, Great Britain; National Heart Lung and Blood Institute, NIH; University of Kansas, Department of Pharmaceutical Chemistry Training Grant, PHS 9/1999; University of Missouri Research Board, 3/2003; Kansas City Area Life Sciences Institute 8/2003.

#### **MANUSCRIPTS REVIEWED**

Nucleic Acids Research; Genomics; The American Journal of Medical Genetics; American Journal of Human Genetics; Journal of Medical Genetics; Clinical Genetics; Cytogenetics and Cell Genetics; Journal of Clinical Endocrinology and Metabolism; Acta Paediatrica; Journal of the American Medical Association; Journal of Pediatric Hematology and Oncology; Human Genetics

#### **INVENTIONS AND PATENTS**

Invention Disclosure 91-1088 (PSU); Detection of Active Genomic Transcription Templates by Synthetic Methylation in Vivo; Disclosure Date: 10/18/91; Status: Disclosed 11/91

Invention Disclosure 92-1151 (PSU); Human Genetic Mapping with Recombinant Disomic Chromosomes; Disclosure Date 6/15/92; Status: Disclosed 11/13/92 - Presented to American Society of Human Genetics

**Invention Disclosure 93- 1226 (PSU) <sup>1</sup>Method for rapid identification of prokaryotic and eukaryotic organisms; US Patent # 5,849,492.**

Invention Disclosure 94- 1339 (PSU); A Method to Define the Chromosomal Location of Disease Genes that Cause Recessive Congenital Disorders; Disclosure Date: 4/18/94

**Invention Disclosure 94- 1440 (PSU/NIH); Computational analysis of nucleic acid information defines binding sites; Status: US Patent # 5,867,402, licensed 3/98**

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<sup>1</sup>Rights to this invention have been acquired by Phylogenetix Laboratories, Inc.

Invention Disclosure 95- 1466 (PSU); 15qllql3 End Clones for Diagnostic Testing of Chromosome Rearrangements and Mutations; Disclosure Date: 4/26/95; Status: Disclosed 10/21/94

Invention Disclosure 95- 1551 (PSU); General Method of Detection of Minimal Residual Disease on Relapse after Non-Autologous Bone Marrow Transplant in Leukemia Patients; Disclosure Date: 12/15/95

Invention Disclosure 97- 0296 (Allegheny General Hospital); Information theory-based analysis of splice junction mutations in hereditary non-polyposis colon cancer; Disclosure Date: 8/1/97; Status: Disclosed 10/24/97

**Invention Disclosure 00-0001 (Children's Mercy Hospital); Selection and generation of single-copy genomic probes for hybridization; Disclosure Date: 3/24/00; Status: Patent application Ser. No. 09/573,080, filed 5/16/00; Allowed 8/13/03; US Patent # 6,828,097 (12/7/04);** Continuation application, filed 2/18/04.

Invention Disclosure 01-0001 (Children's Mercy Hospital); Single copy probes and method of generating same (Continuation-in-part); Disclosure Date: 4/7/00; Status: US Patent App. Ser. No. 09/854,867, filed 5/14/01; PCT/US01/15674.

Invention Disclosure 01-0002 (Children's Mercy Hospital); Subtelomeric DNA probes and method of producing same; Disclosure Date: 9-15-02; Status: US Patent App. Ser. No. 60/415,345, filed 9/30/02. Improved Sub-telomeric DNA Probes and Method of Producing Same, filed 07/2/03. Subtelomeric DNA probes and method of producing same, PCT\US03\31170, WO 2004/029283 A2, US Patent App. Ser # 10\676,248, filed 9/30/03.

Invention Disclosure 01-0004 (Children's Mercy Hospital); Computational selection of probes for localizing chromosome breakpoints in genetic diseases and cancer; Disclosure Date: 4/15/04; Status: US Patent App. Ser #60/557,007, filed 3/26/04.

#### **APPROVED CLINICAL PROTOCOLS**

Study Chair, B957: Genetic Etiology of Acute Leukemia in Children with Down Syndrome, Children's Oncology Group.

Study Committee, C297101: Therapy for Children with Down syndrome and acute leukemia, Children's Oncology Group.

#### **PUBLICATIONS** (in chronological order)

1. Wilson SR, Corvan PJ, Seiders RP, Hodgson DJ, Brookhart M, Hatfield WE, Miller JS, Reiss AH, Rogan PK, Gebert E, Epstein AJ : «The Structure and Magnetic and Electrical Conductivity Properties of the Charge Transfer Compound 1,1-Dimethylferrocenium Bis(tetracyanoquinodimethane),  $(CH_3C_5H_3)_2Fe(TCNQ)_2$ » in *Molecular Metals*, ed. W.H. Hatfield, Plenum Press, p 407-414ff, 1979.
2. Rogan PK, Williams GJB: The structure of the dihydrofolate reductase inhibitor 2,4,6-triamino-5-chloroquinazoline. *Acta Cryst B*36:2358-2362, 1980.
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5. Rogan PK: Restriction mapping by preferential ligation of adjacent digestion fragments. *Nucl Acids Res* 14:9219, 1986.
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8. Rogan PK, Salvo JJ: Molecular genetics of pre-Columbian South American Mummies. In: Molecular Evolution. UCLA Symposium on Molecular and Cellular Biology, 122:223-234, 1990.
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\* Co-principal authors

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37. Saitoh S, Buiting K, Rogan PK, Buxton JL, Driscoll DJ, Arnemann J, Fonig RK, Malcolm S, Horsthemke B, Nicholls RD: Minimal definition of the imprinting center and fixation of a chromosome 15q11-q13 epigenotype by imprinting mutations. *Proc Natl Acad Sci USA* 93:7811-7815, 1996.
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54. Rogan PK, Sabol DW, Punnett H: Maternal uniparental disomy of chromosome 21 in a normal child. Am J Med Genet 83: 69-71, 1999.
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## BOOK CHAPTERS, LETTERS AND CONFERENCE REPORTS

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## **ABSTRACTS**

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1990 Board Certified in Clinical Cytogenetics, American Board of Medical Genetics  
1992 Board Certified in Clinical Cytogenetics, Canadian College of Medical Genetics  
1992 Fellow, Canadian College of Medical Genetics  
1992 Founding Fellow in American College of Medical Genetics  
1996 Board Certified in Clinical Molecular Genetics, American College of Medical Genetics

#### **Academic Appointments:**

1990 Instructor in Pediatrics, Harvard Medical School  
1991 Instructor in Pathology, Harvard Medical School  
1992-99 Assistant Professor in Pathology, Harvard Medical School  
1999- Associate Professor in Medicine/Pediatrics, UMKC School of Medicine

#### **Hospital and Other Appointments:**

1990-99 Research Associate in Medicine (Genetics), Children's Hospital, Boston  
1990-91 Consultant, Integrated Genetics, Framingham, MA  
1991-98 Co-director, Clinical Cytogenetics Service, Beth Israel Deaconess Medical Center, Boston  
1998-99 Director, Clinical Cytogenetics Service, Beth Israel Deaconess Medical Center, Boston  
1999- Director, Clinical Cytogenetics Service, Children's Mercy Hospital, Kansas City  
1999- Director, Human Genetics Research Laboratory, Children's Mercy Hospital, Kansas City  
1999- Professorship in Pediatric Cytogenetics, Children's Mercy Hospital, Kansas City

#### **Committee Assignments:**

##### **National and Regional:**

1991-1995 Scientific Advisor, Angelman Syndrome Foundation  
1992- Scientific Advisor, Canadian Angelman Syndrome Society  
1994- Scientific Advisor, Scientific Advisor, Inverted Duplication 15  
Education and Advocacy Group  
1994- Member, Technology Transfer Subcommittee for diagnostic testing of Angelman and  
Prader-Willi syndromes, American College of Medical Genetics  
1996- Co-organizer of Third International Chromosome 15 Workshop in Vancouver, BC, October 1996  
1999- Member, UM Columbia/Children's Mercy Hospital Genetics Fellowship Joint Training Program

- 2001 Judge, Fellows, Residents & Students Research Presentations, Children's Mercy Hospital's Research Days, June 2001
- 2002-03 Advising Member, Johnson County Community College Biotechnology Steering Committee, Overland Park, KS
- 2003- Member, Basic Sciences Research Committee, Children's Mercy Hospitals, Kansas City

**Memberships and Committee Assignments in Professional Societies:**

- 1986- American Society of Human Genetics
- 1989- Reviewer for Genetics journals (American Journal of Human Genetics, American Journal of Medical Genetics, Cancer Genetics and Cytogenetics, Cytogenetics and Cell Genetics, Genomics, Human Molecular Genetics, New England Journal of Medicine)
- 1991-97 Member, Board of Directors, Angelman Syndrome Foundation
- 1992- Fellow in Canadian College of Medical Genetics
- 1992- Canadian Angelman Syndrome Society
- 1993- Founding Fellow in American College of Medical Genetics
- 1993- Member, Prenatal Diagnosis Committee of New England Regional Genetics Group
- 1993-99 Cytogenetics Faculty Organizer and Member, Harvard Training Fellowship Program in Genetics
- 1994- Member, Inverted Duplication 15 Education and Advocacy Group
- 1999-2004 National Committee for Clinical Laboratory Standards subcommittee advisor on Fluorescence In Situ Hybridization for Medical Genetics for FISH Validation (Report issued 2004)
- 2000, 2001 Laboratory Inspector, College of American Pathologists
- 2001- Active Member, American Cancer Research Association

**Major Research Interests:**

1. Chromatin/chromosomal structure
2. Genetic imprinting of chromosome 15q11q13 in Angelman syndrome, Prader-Willi syndrome, and duplication syndromes
3. Phenotype/genotype relationships in human genetic disorders
4. Molecular mechanisms causing chromosomal rearrangements
5. Bioinformatics and development of new DNA technologies for detection of human genetic disorders

**Principal Clinical and Hospital Service Responsibilities:**

- 1991-1999 Scientific Director, Clinical Cytogenetics Service, Beth Israel Hospital, Boston
- 1999- Director, Clinical Cytogenetics, Children's Mercy Hospital, Kansas City, MO

**Teaching:**

- 1992-99 Genetics Laboratory Tutorials: Demonstrator, first year Harvard medical students
- 1992-99 Cytogenetics lectures, Beth Israel Hospital: Lecturer, first through fourth year Pathology residents
- 1992-99 Cytogenetics Laboratory Rotations: Director of service, students, second year Pathology residents and Genetics Fellows in Harvard Training Program in Genetics, ~5 per year at one month each
- 1993-99 Cytogenetic and molecular genetics lectures for fellows in Harvard Training Program in Genetics
- 1996, 1997 Prenatal Diagnosis Lecture to students in HMS/MIT Reproductive Biology program
- 1999- Management of Continuing Medical Education Lectures at Children's Mercy Hospitals
- 2001- Clinical Cytogenetics, UMKC Medical Students (3<sup>rd</sup> Year)
- 2001- Mentor/Molecular Biology and Cytogenetics Instructor to two Ph.D. Candidates from Kansas University's Department of Chemistry

### **Advising Responsibilities:**

1994 Medical student in research laboratory (Nina Livingston).  
1992-1993 Postdoctoral fellow in research laboratory. (Dr. Sou-De Chang who is currently head of Department of Anatomy. Chang Gung University, College of Medicine, Taipei Taiwan).  
1993-99 Research technicians (Lisa White, Heather Baker).  
1991-99 Clinical technologists (Alena Leff, Min Zhang, Amy Tillman, Shoshana York, JoAnn Rosol-Donoghue, Camille Marsh Scott).  
1992-99 Clinical pathology residents rotating through the clinical laboratory  
1992-99 Faculty, Harvard Genetics Training program  
1995-97 Medical graduate student in research laboratory (Dr. Christine Mundlos).  
1996-98 Clinical genetics fellow in research laboratory (Dr. Gabriella Repetto who is now director of clinical genetics services in University Catolica de Chile, Santiago, Chile).  
1999- 16 clinical laboratory technical staff, 2 office assistants (minimal turnover in last 5 years).  
1999- 2 research laboratory technical staff (Patrick Angell, Mauricio Miralles; previous: Angela Marion, Patricia Walters, Patricia Cazcarro, Brad Dalton), 0.5 office assistant (Ann Lowenstein; previous: Amy Wolfe).

### **Patents/Patent Applications:**

**Invention Disclosure 00-0001 (Children's Mercy Hospital); Selection and generation of single-copy genomic probes for hybridization; Disclosure Date: 3/24/00; Status: Patent application Ser. No. 09/573,080, filed 5/16/00; Allowed 8/13/03; Continuation application, filed 2/18/04. Patent #6,828,097 issued 12/08/04.**

**Invention Disclosure 01-0001 (Children's Mercy Hospital); Single copy probes and method of generating same (Continuation-in-part); Disclosure Date: 4/7/00; Status: US Patent App. Ser. No. 09/854,867, filed 5/14/01; PCT/US01/15674.**

**Invention Disclosure 01-0002 (Children's Mercy Hospital); Subtelomeric DNA probes and method of producing same; Disclosure Date: 9-15-02; Status: US Patent App. Ser. No. 60/415,345, filed 9/30/02. Improved Sub-telomeric DNA Probes and Method of Producing Same, filed 07/2/03. Subtelomeric DNA probes and method of producing same, PCTUS03/31170, WO 2004/029283 A2, US Patent App. Ser # 10/676,248, filed 9/30/03.**

**Invention Disclosure 01-0004 (Children's Mercy Hospital); Computational selection of probes for localizing chromosome breakpoints in genetic diseases and cancer; Disclosure Date: 4/15/04; Status: US Patent App. Ser #60/557,007, filed 3/26/04.**

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#### **Original Reports:**

1. Knoll JHM. Frequency and replication pattern of fragile Xq28 in human heterozygotes from families with X-linked mental retardation. University of Saskatchewan, 1982.
2. Knoll JHM. Roberts Syndrome: Cytological and molecular investigations. University of Manitoba, 1987. (Ph.D. Thesis).
3. Chudley AE, Knoll JH, Gerrard JW, Shepel L, McGahey E, Anderson J. Fragile X-linked mental retardation I: Effect of age and intelligence on expression of the fragile X. *Amer J Hum Genet* 1983; 14:699-712.
4. Knoll JH, Chudley AE, Gerrard JW. Fragile X-linked mental retardation II: Frequency and replication

- pattern of fragile Xq28 in heterozygotes. *Amer J Hum Genet* 1984; 36:640-645.
5. Chudley AE, Rozdilsky B, Houston CS, Becker LE, Knoll JH. Multicore disease in sibs with severe mental retardation, short stature, facial anomalies, hypoplasia of the pituitary fossa, and hypogonadotropic hypogonadism. *Amer J Med Genet* 1985; 20:145-158.
  6. Hagerman RJ, Chudley AE, Knoll JH, Jackson AW, Kemper M, Ahmad R. Autism in fragile X females. *Amer J Med Genet* 1986; 23:375-380.
  7. Heartlein MW, Knoll JHM, Latt SA. Chromosome instability associated with human alphoid DNA transfected into the Chinese hamster genome. *Mol Cell Biol* 1988; 8(9):3611-3618.
  8. Knoll JHM, Nicholls RD, Magenis E, Graham JM Jr, Lalande M, Latt SA. Angelman and Prader-Willi syndromes share a common chromosome 15 deletion but differ in parental origin of the deletion. *Amer J Med Genet* 1989; 32(2):285-290.
  9. Nicholls RD, Knoll JH, Glatt K, Hersh JH, Brewster TD, Graham JM Jr, Wurster-Hill D, Wharton R, Latt SA. Restriction fragment length polymorphisms within proximal 15q and their use in molecular cytogenetics and the Prader-Willi syndrome. *Amer J Med Genet* 1989; 33:66-77.
  10. Knoll JHM, Nicholls RD, Lalande M. On the parental origin of chromosome 15q11q13 in Angelman Syndrome. *Hum Genet* 1989; 83:205-206.
  11. Nicholls RD, Knoll JHM, Butler MG, Karam S, Lalande M. Genetic imprinting suggested by maternal heterodisomy in nondeletion Prader-Willi syndrome. *Nature* 1989; 342:281-285.
  12. Knoll JHM, Nicholls RD, Magenis RE, Glatt K, Graham JM Jr, Kaplan L, Lalande M. Angelman Syndrome: Three molecular classes identified with chromosome 15q11q13 specific DNA markers. *Amer J Hum Genet* 1990; 47:149-155.
  13. Bianchi DW, Flint AF, Pizzimenti MF, Knoll JHM, Latt SA. Isolation of fetal DNA from nucleated erythrocytes in maternal blood. *Proc Nat Acad Sci* 1990; 87(9):3279-3283.
  14. Knoll JHM, Glatt K, Nicholls RD, Malcolm S, Lalande M. Chromosome 15 uniparental disomy is not frequent in Angelman Syndrome. *Amer J Hum Genet* 1991; 48:16-21.
  15. Wagstaff J, Knoll JHM, Fleming J, Kirkness EF, Martin-Gallardo A, Greenberg F, Graham JM Jr, Menninger J, Ward D, Venter JC, Lalande M. Localization of the gene encoding the GABA<sub>A</sub> receptor B3 subunit to the Angelman/Prader-Willi region of human chromosome 15. *Amer J Hum Genet* 1991; 49:330-337.
  16. Chaillet JR, Knoll JHM, Horsthemke B, Lalande M. The syntenic relationship between the critical deletion region for the Prader-Willi/Angelman syndromes and proximal mouse chromosome 7. *Genomics* 1991; 11:773-776.
  17. Buiting K, Greger V, Horstmann I, Ludecke JJ, Senger G, Claussen U, Brownstein BH, Schlessinger D, Knoll JHM, Lalande M, Zabel B, Horsthemke B. Microdissection and molecular analysis of proximal 15q. In: Cassidy SB, ed. *Prader-Willi syndrome and other chromosome 15q deletion disorders*. Springer Verlag, Berlin. Series H: Cell Biology 1991; 61:13-17.

18. Beggs AH, Byers TJ, Knoll JHM, Boyce FM, Bruns G, Kunkel LM. Cloning and characterization of two human skeletal muscle alpha-actinin genes on chromosomes one and eleven. *J Biol Chem* 1992; 267:9281-9288.
19. Wagstaff J, Knoll JHM, Glatt KA, Shugart YY, Sommer A, Lalande M. Linkage of nondeletion Angelman syndrome to chromosome 1511-q13: Maternal but not paternal transmission leads to phenotypic expression. *Nature Genetics* 1992; 1:291-294.
20. Webb T, Clayton-Smith J, Cheng X-J, Knoll JHM, Lalande M, Pembrey ME, Malcolm S. Angelman syndrome with a chromosomal inversion 15 (p11q13) accompanied by a deletion in 15q11q13. *J Med Genet* 1992; 29:921-924.
21. Knoll JHM, Wagstaff J, Lalande M. Cytogenetic and molecular studies in Prader-Willi and Angelman syndromes. *Amer J Med Genet* 1993; 17:694-698.
22. Knoll JHM, Sinnott D, Wagstaff J, Glatt K, Wilcox AS, Whiting P, Wingrove P, Sikela JM, Lalande M. FISH ordering of reference markers and of the gene for the  $\alpha 5$  subunit of the gamma-aminobutyric acid receptor (GABRA5) within the Angelman and Prader-Willi syndrome chromosome regions. *Hum Molec Genet* 1993; 2:183-189.
23. Warman ML, Tiller GE, Polumbo PA, Seldin MF, Rochelle JM, Knoll JHM, Cheng SD, Olsen BR. Physical and linkage mapping of the human and murine genes for the  $\alpha 1$  chain of type IX collagen (COL9A1). *Genomics* 1993; 17:694-698.
24. Peters K, Knoll JHM. Diagnosis of tumors: The application of cytogenetics and fluorescence-in-situ-hybridization. *Verh Dtsch Ges Zyt* 1993; 18:66-68.
25. Oh SP, Warman P, Selden M, Cheng SD, Knoll JHM, Timmons S, Olsen BR. Cloning of cDNA and genomic DNA encoding human type XVIII collagen and localization of the  $\alpha 1$ (XVIII) collagen gene to mouse chromosome 10 and human chromosome 21. *Genomics* 1994; 19: 494-499.
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29. Cheng SD, Spinner NB, Zackai EH, Knoll JHM. Cytogenetic and molecular characterization inverted duplicated chromosomes 15 from eleven patients. *Amer J Hum Genet* 1994; 55:753-759.
30. Knoll JHM, Asamoah A, Pletcher BA, Wagstaff J. Interstitial duplication of proximal 22q: Phenotypic overlap with cat eye syndrome. *Amer J Med Genet* 1995; 55:221-224.
31. Mundlos S, Mulliken JB, Abramson DL, Warman ML, Knoll JHM, Olsen BR. Genetic mapping of cleidocranial dysplasia and evidence of a microdeletion in one family. *Hum Mole Genet* 1995; 4:71-75.

32. White L, Knoll JHM. Angelman syndrome: Routine molecular cytogenetic analysis of chromosome 15q11-q13. *Amer J Med Genet* 1995; 56:101-105.
33. Spinner NB, Zackai E, Cheng SD, Knoll JHM. Supernumerary inv dup(15) in a patient with Angelman syndrome and a deletion of 15q11-q13. *Amer J Med Genet* 1995; 57:61-65.
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35. Greger V, Knoll JHM, Woolf E, Glatt K, Tyndale RF, Olsen RS, Tobin AJ, Sikela JM, Nakatsu Y, Brilliant M, Whiting PJ, Lalande M. The  $\tau$ -aminobutyric acid receptor  $\tau 3$  subunit gene (GABRG3) is tightly linked to the  $\alpha 5$  subunit gene (GABRA5) on human chromosome 15q11-q13 and is transcribed in the same orientation. *Genomics* 1995; 26:258-264.
36. Azim AC, Knoll JHM, Beggs AH, Chishti AH. Isoform cloning, actin binding, and chromosomal localization of human erythroid dematin, a member of the villin superfamily. *J Biol Chem* 1995; 270:17404-17413.
37. Simmons CF, Jr, Clancy TE, Quan R, Knoll JHM. The oxytocin receptor gene (OXTR) localizes to human chromosome 3p25 by fluorescence in situ hybridization and PCR analysis of somatic cell hybrids. *Genomics* 1995; 26:623-625.
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39. Schwartz F, Eisenman R, Knoll J, Gessler M, Bruns G. cDNA sequence, genomic organization and evolutionary conservation of a novel gene from the WAGR region. *Genomics* 1995; 29(2):526-532.
40. Azim AC, Knoll JHM, Marfatia SM, Peel DJ, Bryant PJ, Chishti AH. hDlg: Chromosome location of the closest human homologue of the Drosophila discs large tumor suppressor gene. *Genomics* 1996; in press.
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43. White L, Rogan PK, Nicholls RD, Wu B-L, Korf B, Knoll JHM. Allele-specific replication of 15q11q13 loci: A diagnostic test for detection of uniparental disomy. *Amer J Hum Genet* 1996; 59:423-430.
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60:574-580.

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51. Repetto GR, Korf BR, Wagstaff J, Knoll JHM. Complex familial rearrangement of chromosome 9p24 detected by FISH. *Amer J Med Genet*, 1998; 76:306-309.
52. Blank V, Knoll JHM, Andrews NC. Molecular characterization and localization of the human MAFG gene. *Genomics* 1997; 44:147-149.
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55. Ott G, Katzenberger T, Siebert R, DeCoteau JF, Fletcher JA, Knoll JHM, Kalla J, Rosenwald A, Michaelson M, Ott MM, Weber-Matthiesen K, Kadin ME, Muller-Hermelink HK. Chromosomal abnormalities in nodal and extranodal CD30+ anaplastic large cell lymphomas: Infrequent detection of the t(2;5) in extranodal lymphomas. *Genes, Chromosomes and Cancer* 1998; 22:114-121.
56. Rogan PK, Seip JR, White LM, Wenger SL, Steele MW, Sperling MA, Menon R, Knoll JHM. Relaxation of imprinting in Prader-Willi syndrome. *Human Genetics* 1998; 103:694-701.
57. Sterpetti P, Hack A, Bashir M, Cheng SD, Knoll JHM, Toksoz D. Activation of the Lbc Rho Exchange Factor-Proto-oncogene by truncation of an extended C-terminus that regulates transformation and targeting. *Mol Cell Bio* 1999; 19(2); 1334-1345.
58. Repetto GM, White LM, Bader PJ, Johnson D, Knoll JHM. Interstitial duplication of proximal chromosome 15q in three patients with autistic features and mental retardation. *Amer J Med Genet* 1998; 79:82-89.

59. Hahn WC, Stewart SA, Brooks M, York SG, Ng-Eaton E, Kurachi A, Beijersbergen RL, Knoll JHM, Meyerson M, Weinberg RA. Inhibition of telomerase limits the growth of human cancer cells. *Nature Medicine* 1999; 5:1164-1170.
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61. Austin-Ward ED, Castillo S, Gragnic Y, Sanz P, Salazar S, Knoll JHM. Clinical findings in a patient with a supernumerary ring chromosome 20. *Amer J Med Genet* 91:171-174, 2000.
62. Ming JE, Blagowidow N, Knoll JH, Rollings L, Fortina P, McDonald-McGinn DM, Spinner NB, Zackai EH. A submicroscopic deletion in cousins with Prader-Willi syndrome causes a grandmatrilineal inheritance pattern: effects of imprinting. *Amer J Med Genet* 92:19-24, 2000.
63. Rogan, PK, Cazcarro, P, Knoll JHM. Sequence-based design of single-copy genomic DNA probes for fluorescence in situ hybridization. *Genome Research* 11: 1086-1094, 2001.
64. O'Sullivan, MJ, Swanson, PE, Knoll, J, Taboada, EM, Dehner, LP. Undifferentiated embryonal sarcoma with unusual features arising within mesenchymal hemartoma of the liver: Report of a case and review of the literature. *Pediatr Dev Pathol*. Sep-Oct 4(5): 482-489, 2001.
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66. Knoll JHM, Rogan PK. Sequence-based, *in situ* detection of chromosomal abnormalities at high resolution, *American Journal of Medical Genetics*, 121A: 245-257, 2003.
67. Rogan PK, Knoll JHM. High resolution detection of chromosome abnormalities with single copy fluorescence in situ hybridization. *IEEE Symposium on Biomedical Engineering*, April, 2004.
68. Rogan PK, Knoll JHM: High Resolution Definition of Chromosome Abnormalities with Probes Designed from Genome Sequences, *Discovery Medicine*, 21(4): 99-101, 2004.
69. Tetsuya K, Shyu C, He L, Rogan PK, Knoll JHM. Content and classification based ranking algorithm for metaphase chromosome images. *Proceedings of the IEEE International Conference*, accepted for publication.
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71. Knoll JHM, Angell P, Walters P, Marsh C, Rogan P. Generation of high-specificity, single-copy probes proximate to human telomeres. Submitted 2004.

### **Reviews and educationally relevant publications:**

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3. Lalande M, Wagstaff J, Sinnett D, Greger V, Knoll JHM. Mapping of the Angelman and Prader-Willi syndromes. In Epstein CJ (ed): The phenotypic mapping of Down syndrome and other aneuploid conditions, 1993; 225-234, Wiley-Liss, New York.
4. Knoll JHM, Lichter P. In situ hybridization to metaphase chromosomes and interphase nuclei. In Dracopoli NC, Haines JL, Korf BR, Moir DT, Morton CC, Seidman CE, Seidman JG, Smith DR (eds): "Current protocols in Human Genetics Volume 1" 1994: Unit 4.3, Green-Wiley, New York. (Revised July, 2004).
5. Knoll JHM. Prader-Willi Syndrome. In: Smith B, Adelman G, Eds. Neuroscience Year: Supplement 4 to the *Encyclopedia of Neuroscience*. Birkhauser, Boston, MA. 1996.
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7. Knoll JHM. Genomic Imprinting. In: McGraw-Hill 1999 Yearbook of Science and Technology. December, 1998.
8. Knoll JHM. A Cytogenetic nightmare. Case 4 for Genetics, Embryology, and Reproduction for first year medical students, Harvard Medical School, 1997; 1998; 1999.
9. Knoll JHM, Rogan PK: Prader Willi Syndrome, Encyclopedia of Neuroscience, 3<sup>rd</sup> edition (George Adelman and Barry H. Smith, eds.), in press.

### **Abstracts (last 5 years):**

1. The human SRA1 gene maps proximal to the Prader-Willi/Angelman syndrome domain in 15q11 and is non-imprinted. *Amer J Hum Genet* 65(4):A458, 1999.
2. JHM Knoll, H Baker, G Cox, ML Begleiter, LM Pasztor. Parental origin and replication timing studies in a 70, XXXX liveborn. *Amer J Hum Genet* 65(4):A167, 1999.
3. Kruskall MS, Uhl L, Yu N, Hussein S, Yunis E, Knoll J. Misinterpretation of HLA typing in a woman with congenital chimerism due to postzygotic fusion of two embryos. American Association of Blood Banking Meeting, November, 2000.
4. Knoll JHM, Cazcarro P, Rogan PK: Clinical application of sequence-based single copy probes for FISH, American Society of Human Genetics, October 2000.
5. Rogan PK, Cazcarro P, Knoll JHM. Single copy hybridization probes derived by genomic sequence analysis. American Society of Human Genetics, October 2000

6. Rogan PK, Cazcarro P, Knoll JHM: Single copy hybridization probes for detection of chromosome rearrangements derived by genomic sequence analysis. American Association for Cancer Research, March 2001.
7. Rogan PK, Cazcarro P, Knoll JHM: Single copy hybridization probes for detection of chromosome rearrangements derived by genomic sequence analysis. 10<sup>th</sup> International Congress of Human Genetics (platform), May 2001.
8. Rogan PK, Cazcarro P, Knoll JHM: Novel features of genome organization revealed by single copy FISH. American Society of Human Genetics, October 2001.
9. Cazcarro P, Rogan PK, Knoll JHM: Rapid sequence-based definition of chromosomal abnormalities. American Society of Human Genetics, October 2001.
10. Knoll JHM, Rogan PK: Detection of chromosomal rearrangements with sequence-defined, single copy hybridization probes. Second International NCI-EORTC Meeting on Cancer Diagnostics: From Discovery to Clinical Practice. June, 2002.
11. Knoll JHM, Angell P, Rogan PK: Detection of chromosomal rearrangements with single copy FISH probe arrays. American Society of Human Genetics, October 2002.
12. Knoll JHM, Rogan PK. Detection of Chromosomal Rearrangements with Single Copy FISH. 2<sup>nd</sup> Annual Kansas City Area Life Sciences Day, March 2003.
13. J.H.M. Knoll & P.K. Rogan. Detection of Chromosomal Rearrangements with Single Copy FISH. XIX International Congress of Genetics, July 2003.
14. Joan H.M. Knoll and Peter Rogan. Personalized Molecular Cytogenetic Diagnosis. 3<sup>rd</sup> Annual Kansas City Area Life Sciences Day, June 2004.
15. Knoll JHM, Marion AM, Fletjer W, Persons D, Cowan J, Rogan PK. Small upstream deletions of *ABL* 1 are rare in Chronic Myelogenous Leukemia. 2004 ASHG Annual Meeting, November, 2004.
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